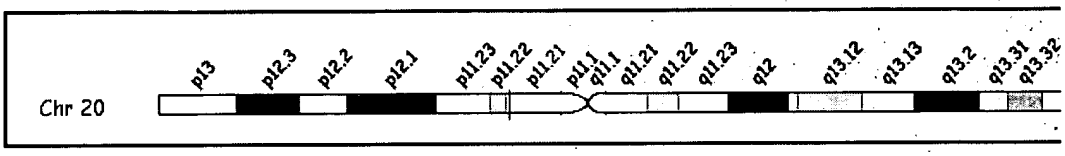
GeneCards™ an academic web site of the WEIZMANN INSTITUTE OF SCIENCE

GeneCard for gene THBD

Approved UCL/HGNC/HUGO Human Gene Nomenclature database symbol
THBD (thrombomodulin)

Aliases and Additional Descriptions (According to GDB, HUGO, and/or SWISS-PROT)	• THRM • **thrombomodulin** • **Thrombomodulin** precursor (Fetomodulin) (TM) (CD141 antigen).
Chromosomal Location (According to LocusLink and/or UDB and/or HUGO, Genomic Views According to UCSC and Ensembl)	*Chromosome:* **20** *LocusLink cytogenetic band:* **20p12-cen** *Ensembl cytogenetic band:*  *Unified DataBase coordinate (from pter):* 24,473 mega bases *Genomic View:* UCSC Golden Path
Proteins (According to SWISS-PROT and/or MIPS)	TRBM_HUMAN **Size**: 575 amino acids; 60329 Da **Function: thrombomodulin is a specific endothelial cell receptor that forms a 1**: 1 ST CONVERSION OF PROTEIN C TO THE ACTIVATED PROTEIN C (PROTEIN CA). ONCI MECHANISM, FACTOR VA AND FACTOR VIIIA, AND THEREBY REDUCES THE AMOU **Subcellular location**: Type I membrane protein. **Tissue specificity**: ENDOTHELIAL CELLS ARE UNIQUE IN SYNTHESIZING **THROMBO** **Polymorphism**: VARIATIONS IN THBD ARE ASSOCIATED WITH AN INCREASED RISI **Similarity**: CONTAINS 6 EGF-LIKE DOMAINS. **3D structures:** PDB ids 1EGT (3D) 1FGD (3D) 1FGE (3D) 1TMR (3D) 1ZAQ (3D) **MIPS Pedant Viewer:** 682 **REFSEQ proteins:** NP_000352.1
Protein Domains/Families (According to BLOCKS and/or InterPro)	**Blocks protein families:** BL00615 C-type lectin domain proteins. BL01187 Calcium-binding EGF-like domain proteins pattern proteins. PR00907 Thrombomodulin signature **InterPr Domains and Families:** IPR001304; Lectin_C IPR001491; Thrmbomoduln

IPR000561; EGF-like
IPR001881; EGF_Ca
IPR000152; Asx_hydroxyl

Graphical View of Domain Structure for SP Entry P07204

Sequences (GenBank/EMBL/DDBJ Accessions According to Unigene or GenBank, RefSeq According to LocusLink, Assembly According to MIPS and/or DOTS)

REFSEQ mRNAs: NM_000361.1

Additional Gene/cDNA sequence:
D00210.1 J02973.1 M16552 M16552.1 X05495 X05495.1

MIPS assembly:H426S1

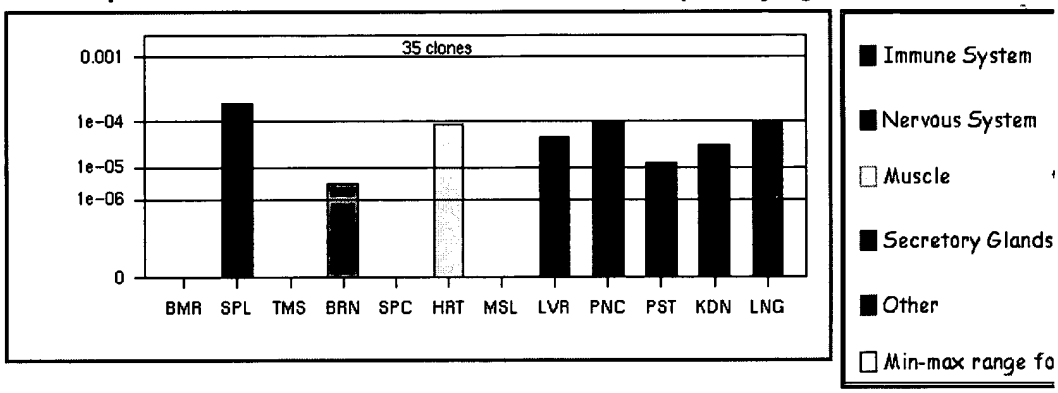
DOTS assembly:
DT.416446 DT.92427530 DT.92427529

Unigene Cluster for THBD: (Build 151 Homo sapiens; May 27 2002)
thrombomodulin
Hs.2030 [show with all **ESTs**]

Unigene Representative Sequence: NM_000361

Expression in Human Tissues (According to proprietary W.I.S DNA array results, UniGene and/or SOURCE)

THBD expression in normal human tissues based on quantifying ESTs from vari us



SOURCE GeneReport for Unigene cluster Hs.2030

Similar Genes in Other Organisms (According to MGD Jun 10 2002 , Stony Brook C.elegans-H.sapiens Alignment Database and/or euGenes)

Homologues:

	gene	*locus*	*description*
mouse (MGD)	Thbd	2 (84.00 cM)	thrombomodulin
fly (euGenes)	ple	3 65C3	catecholamine metabolism tyrosine 3-monooxygen
C. elegans (Stony Brook)	W07G4.4	--	description: ke58e03.y1 Dirofilaria immitis adult SL immitis cDNA similar to SW:YH24_CAEEL Q27245 AMINOPEPTIDASE W07G4.4 IN CHROMOSOME

Variants: SWISS-PROT: TRBM_HUMAN

NCBI SNPs: 10/18 selected, not withdrawn, single nucleotide mutations are shown here.
Click here to see all of them

SNPs/Variants (According to theNCBI SNP Database and to SWISS-PROT)

Genomic Data					
SNP ID	Contig Accession	Position in Contig	Strand	5' Flanking Sequence*	3' Flanking Seque
rs1042579	NT_011387.7	22966781	-	CCGACTCGGCCCTTG	CCGCCACATTGC
rs3176121	NT_011387.7	22966313	-	CTAACTGGCGAGGGG	TGATTAGAGGGA
rs3176122	NT_011387.7	22965974	-	GTAAACTATCTTGGT	AATTTTTTTTTC
rs3176123	NT_011387.7	22965470	-	GGTTGCTCTAGATTG	GAGAAGAGACA
rs3176124	NT_011387.7	22965002	-	TCAGGCCCTTATTTT	AAGAAACTGAGC
rs3176133	NT_011387.7	22966384	-	CACCTTAGCTGGCAT	ACAGCTGGAGA
rs3176134	NT_011387.7	22966194	-	CAGGTCCTCACTACC	GGCGCAGGAGG
rs1042580	NT_011387.7	22965678	-	TGAGATGTAAAAGGT	TTAAATTGATGT
rs3176117	NT_011387.7	22969818	-	GACGCCATACTCTCT	TTCTTGTTTAAA
rs3176119	NT_011387.7	22969167	-	CAATTCACCTGCCAC	GCCTCTGAGCCC

** Lower case letters indicate repetitive or low-complexity sequence*

All NCBI SNPs in THBD

Disorders & Mutations (in which this Gene is Involved, According to OMIM, SWISS-PROT, Genatlas, GeneClinics, HGMD, BCGD, and/or TGDB.)

OMIM ID: 188040

search databases for MIM named disorders:
- Thrombophilia due to **thrombomodulin** defect
- {Myocardial

SWISS-PROT: TRBM_HUMAN

- **Disease**: DEFECTS IN THBD COULD BE THE CAUSE OF INHERITED TED, ALSC HAEMOPOIETIC SYSTEM WHICH CREATES A TENDENCY TO THE OCCURREN CARDIOVASCULAR DISORDERS.

Genatlas disease: THBD

- thrombosis,recurrent

Human Gene Mutation Database entry for THBD

Medical News (Possibly Related Articles in Doctor's Guide)

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Research Articles (in PubMed)

- Structure and expression of human **thrombomodulin**, a thrombin receptor on endotl

Search PubMed for THBD to find abstracts of **research articles** containing

THBD in Other Genome Wide Resources: (According to GDB, LocusLink, euGenes, Ensembl and/or GeneLynx)

GDB: 119613 LocusLink: 7056 euGenes: HUgn7056 Ensembl: ENSG0000010

THBD in General Databases, Limited Scope (According to HUGE)

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THBD in Specialized Databases (According to ATLAS, GENATLAS, HORDE, IMGT, MTDB and/or SWISS-PROT)	*name* **G natlas** biochemistry entry for THBD: thrombomodulin coagulation factor complexing w mutations in the promoter region putatively associated with a risk for arterial thrombosis a PROW -CD guide CD141 entry.
Services (According to RZPD)	Search RZPD for clones of THBD Clone collection at the German Human Genome Proje

Back (to Search Results) - **More** like this

Search the web for THBD - search millions of **Web pages** with **Excite** to find other web sites related

GeneCards **Homepage** - How to **Search** or **Cite** this Database - Last **Update**: 20 Jun 2002

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Display the GeneCard of a random gene
Display the GeneCard of a random HUGO-approved gene

The GeneCards **idea** in brief: **Mining** the Internet for biomedical knowledge and **guiding** the user to it.

Developed at the Crown Human Genome Center & Weizmann Institute of Science

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